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Rectal Atresia as Rare Manifestation in EEC Syndrome

F. Majewski and T. Goecke
Institute of Human Genetics, Düsseldorf, Germany

A newborn boy presented with bilateral split hand/foot malformation, sparse hair, dry and scaly skin, and nasolacrimal duct obstruction. Despite absence of cleft lip or palate, the findings fit the EEC syndrome. Additionally, the boy had rectal atresia. At least six further patients with EEC syndrome and anal atresia (two published, four unpublished) demonstrate that anorectal malformation is a further, but rare anomaly in EEC syndrome. © 1996 Wiley-Liss, Inc.

KEY WORDS: EEC syndrome, anal/rectal atresia

INTRODUCTION

The EEC syndrome is characterized by ectrodactyly (split hand/split foot, syndactyly), ectodermal dysplasia (anomalies of hair, teeth, nails, nasolacrimal ducts) and cleft lip/palate. More than 100 patients have been described. This syndrome is an autosomal dominant trait with reduced penetrance and a wide variability of expression. Küster et al. [1985], Majewski et al. [1988], and Fryns et al. [1990] demonstrated that each manifestation can be absent in affected patients. There are some further anomalies, as deafness [Buss et al.,1995], choanal atresia, renal anomalies, and anal atresia [de Smet and Fryns, 1995]. We report on an affected newborn infant with EE(C) syndrome and intermediate rectal atresia.

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Address reprint requests to Prof. Dr. F. Majewski, Institut für Humangenetik, Moorenstrasse 5, 40225 Düsseldorf, Germany.

Dedicated to Jürgen W. Spranger on the occasion of his 65th birthday with admiration and best wishes.

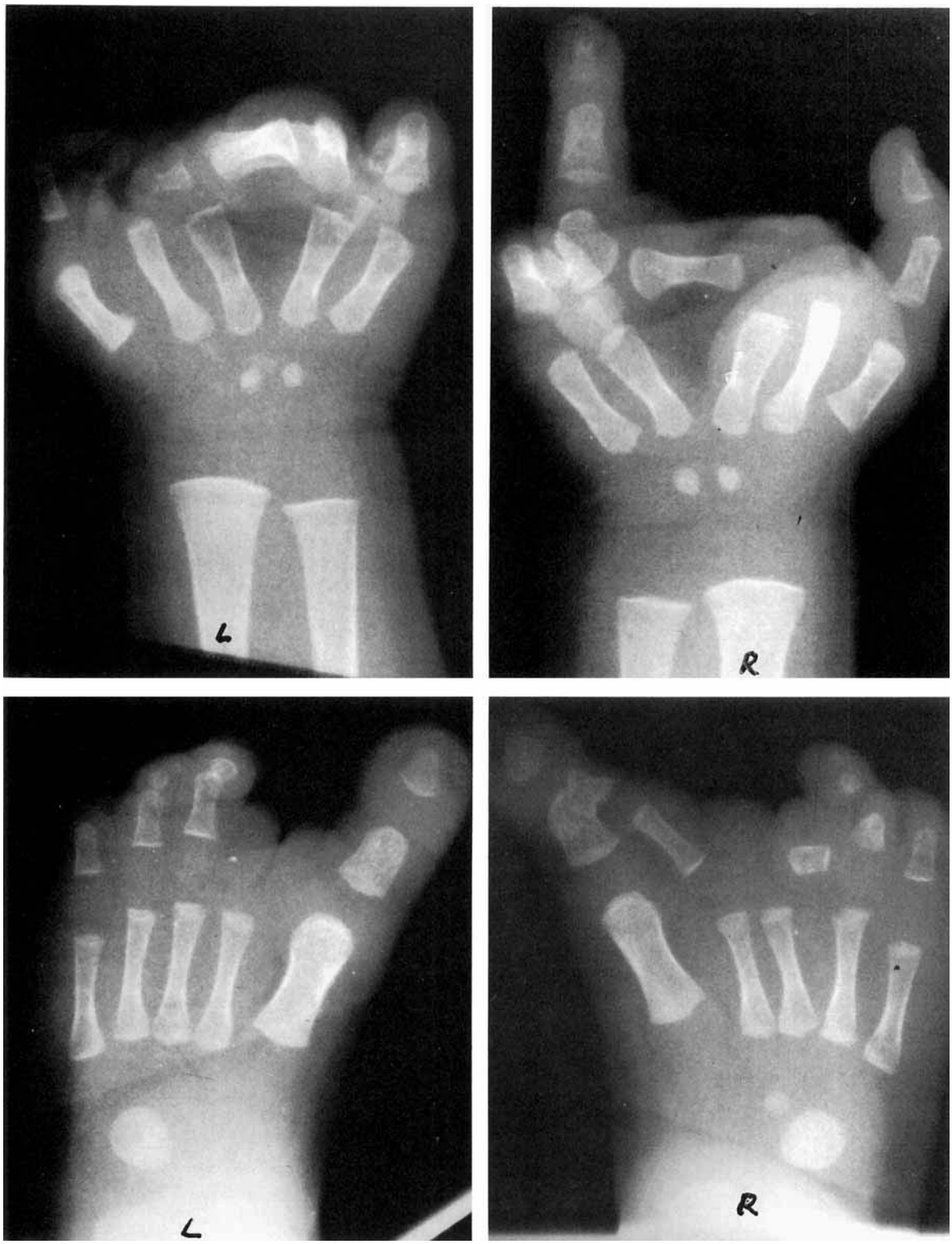

Fig. 2. Roentgenogram of the hands and feet demonstrating typical split hands and split feet.

CLINICAL REPORT

This newborn boy is the first child of healthy, unrelated parents. All other relatives are reported to be normal. Whereas the father appeared normal, the mother had thin hair, marked caries, labial displacement of the right upper canine, high root and full tip of the nose, and brittle finger nails.

After an uneventful pregnancy the baby was born spontaneously in the 38th week of gestation. Birth weight was 2,820 g, length was 50 cm, and OFC was 35 cm. The boy had bilateral split hands and feet and intermediate rectal atresia. Anus and sphincter ani were present. Rectal atresia was treated surgically.

We examined the boy at age 10 weeks. Length was 57 cm, weight was 4,840 g, and OFC was 37 cm. The occiput was elongated, scalp hair was sparse, and skin of the scalp, forehead, and face was dry and scaly. The eyebrows were nearly absent; the eyelashes sparse. The eyelids were inflamed. The tip of the nose was somewhat bulbous, there was a median raphé of the philtrum, and the palate was closed. There was an artificial anus and rectal atresia just above the sphincter ani. The boy appeared neurologically normal. Both arms and legs were normal except a bilateral split hand and foot malformation (Figs. 1, 2).

On the right hand, the thumb and 5th finger were normal, but there was camptodactyly of the 4th finger. The second and third fingers were absent. All metacarpals were present, and an additional transverse bone was palpable distally to metacarpals 2–4. Nails 1, 4, and 5 appeared normal. On the left hand, the thumb and thenar eminence was hypoplastic, the small distal phalanx was contract. Fingers 2 and 3 were absent, the 4th finger was contracted and deviated medially, and finger 5 was normal. All metacarpals were present, and there was the same transverse bone as on the right hand. The nail of the thumb was small; the nails of the fingers 4 and 5 were normal. On the right foot, the second toe was absent, and there was complete cutaneous syndactyly of toes 3 and 4. All nails were normal. On the left foot, the hallux deviated medially, the first and second toes were syndactylous, the third toe was absent, and all metatarsals were present with an additional transverse bone going from metatarsal 3 to metatarsal 4.

X-ray examination (Fig. 2) showed the malformations as described above. There were transverse bones in both hands corresponding to metacarpals 3 and 4, which deviated like a V distally, indicating a cleft. In the right foot there was osseous syndactyly of the first and second toe, and a transverse bone corresponding to the metatarsals 3 and 4.

DISCUSSION

The EEC syndrome is an autosomal dominant trait with reduced penetrance and highly variable expression. As shown by affected families none of the three major malformations, e.g., ectrodactyly, ectodermal dysplasia, and clefting, is obligate [Küster and Majewski, 1985; Majewski and Küster, 1988; Fryns et al., 1990]. Since our patient had ectodermal dysplasia (sparse hair of the scalp, sparse eyebrows, dry and scaly skin, obliteration of the nasolabial ducts), and tetramelic split hand/foot malformation, we think the diagnosis EEC syndrome (without clefting) is correct.

Whether the mild anomalies of the mother, e.g., the somewhat bulbous tip of the nose [described by de Smet and Fryns, 1995, as typical in a mildly affected mother], the caries, and the thin scalp hair are symptoms of the EEC syndrome, may be speculative unless a molecular analysis of the gene is possible. Since at least one typically affected three generation family exhibited a balanced reciprocal translocation between 7q11.21 and 9p12 [Hasegawa et al., 1991], and the gene for isolated split hand/foot is thought to be located at 7q21.2-7q22.1 [Genuardi et al., 1994], the EEC-gene may be located at 7q11-7q22.

Apart from the three major malformations, additional anomalies have been described: conductive deafness, choanal atresia, and urogenital anomalies, especially hypospadias and hydronephrosis [Buss et al., 1995].

Anorectal malformations were reported in a few EEC cases. Rosenmann et al. [1976] first reported imperforate anus in a typically affected boy. The father of this boy was typically affected too, but without anal atresia. De Smet and Fryns [1995] reported on a newborn girl with EEC syndrome, omphalocele, and anal atresia. The mother was mildly affected and without anorectal anomalies. An additional literature survey showed no further reports on similarly affected patients with the EEC syndrome. However, since at least four unpublished patients with EEC syndrome have anal atresia [A. Schinzel, N. Le Merrer, quoted by de Smet and Fryns, 1995], it seems likely that the association of EEC syndrome and anorectal malformation is non-random.

REFERENCES

Buss PW, Hughes HE, Clarke A (1995): Twenty-four cases of the EEC syndrome: clinical presentation and management. J Med Genet 26:586–589.

De Smet L, Fryns JP (1995): Anal atresia and abdominal wall defect as unusual symptoms in EEC syndrome. Genet Counsel 6:127–130.

Fryns JP, Legius E, Dereymaeker AM, van den Berghe H (1990): EEC syndrome without ectrodactyly: Report of two new families. J Med Genet 27:165–168.

Genuardi M, Gurrieri F, Neri G (1994): Genes for split hand/split foot and laterality on 7q22.1 and Xq24–q27.1. Am J Med Genet 50:101.

Hasegawa T, Hasegawa Y, Asamura S, Nagai T, Tsuchiya Y, Ninomiya M, Fukushima Y (1991): EEC syndrome (ectrodactyly, ectodermal dysplasia and cleft lip/palate) with a balanced reciprocal translocation between 7q11.21 and 9p12 (or 7p11.2 and 9q12) in three generations. Clin Genet 40:202–206.

Küster W, Majewski F, Meinecke P (1985): EEC syndrome without ectrodactyly? Report of 8 cases. Clin Genet 28:130–135.

Majewski F, Küster W (1988): EEC syndrome sine sine? Report of a family with oligosymptomatic EEC syndrome. Clin Genet 33:69–73.

Rosenman A, Shapira T, Cohen MM (1976): Ectrodactyly, ectodermal dysplasia and cleft palate (EEC syndrome). Report of a family and review of the literature. Clin Genet 9:347–353.